Diseases prove elusive in global genetic search

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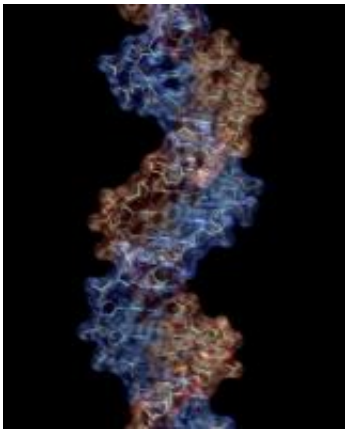

A rendered image of DNA.

(PhysOrg.com) -- A failure to use large international sample groups when searching for the genetic basis of common diseases is contributing to a lack of knowledge about the true frequency of illnesses across populations, according to an Australian National University researcher.

Professor Julio Licinio, the Director of The John Curtin School of Medical Research at ANU, is one of a team of researchers analysing data from the Human Haplotype Mapping (HapMap) project. In a new study, published today in *Nature*, the researchers reveal the results of a study that searched for a map of genetic variations that may be the basis of disease across a broad sample from around the world.

They found that even when drawing on large sample groups, that there was a high frequency of rare genetic variants, which will make it necessary to study large numbers of people in order to better understand the genetics of common disorders. This suggests, they say, that is better to simply start afresh with new genetic research, rather than use that which is based on small sample groups.

“The results from looking at this broad international group has shown that statistical correlations between rare genetic variants discovered by sequencing and common sequence variations is low and even their imputation can be practically difficult when the reference panel isn’t large enough,” said Professor Licinio.

“These data point to a much greater use for de novo sequencing approaches - in effect starting afresh - for the discovery of rare variants that contribute to human phenotypes, than can be realised by strategies that test for variants discovered in sample sets that are modest in size or drawn from different populations than are to be tested.”

The research drew on the data from a wide range of populations around the world - from Africa to the USA and Europe.

“In contrast to previous studies, this used a broad sample of human populations including Yoruba from Ibadan, Nigeria, Americans of north European descent in Utah, Han Chinese from Beijing in China, Maasai from Kenya, Toscani in Italy, Mexican-Americans, and many more.

“The results provide for the first time a framework of the properties and parameters of rare and complex diseases in humans’ genetic code, contrasted with well-known illnesses,” said Professor Licinio. “This study serves as an important step towards a high resolution landscape of the human genome.”